GENETICS

WHAT IS GENETICS?

Genetics is the study of heredity i.e., transmission of body features (both similarities and differences) from parents to offspring and the laws relating to such transmission.

GREGOR MENDEL - An Austrian Monk and Father of Genetics

Gregor Mendel (1822 - 84) was born in a peasant family. He had his early education in a monastery and later he studied Science and Mathematics at University of Vienna. He wanted to be a teacher but luck did not favour him, and he failed in the examination of teaching certificate. His findings are now called as "Mendel's laws of inheritance".

Two modern applications of genetics

Genetic engineering is the technique in which the genetic constitution of an organism (bacterium) is altered by introducing new genes into its chromosomes.

Genetic Counselling - Diseases like haemophilia (bleeder's disease with a tendency to bleed freely from even a slight wound), thalassaemia and sickle cell anaemia with defective haemoglobin are examples of genetic diseases which can be prevented to some extent by proper genetic matching of the prospective parents.

Heredity

The term heredity may be defined as "transmission of genetically based characteristics from parents to offspring", or "the genetic constitution of an individual".

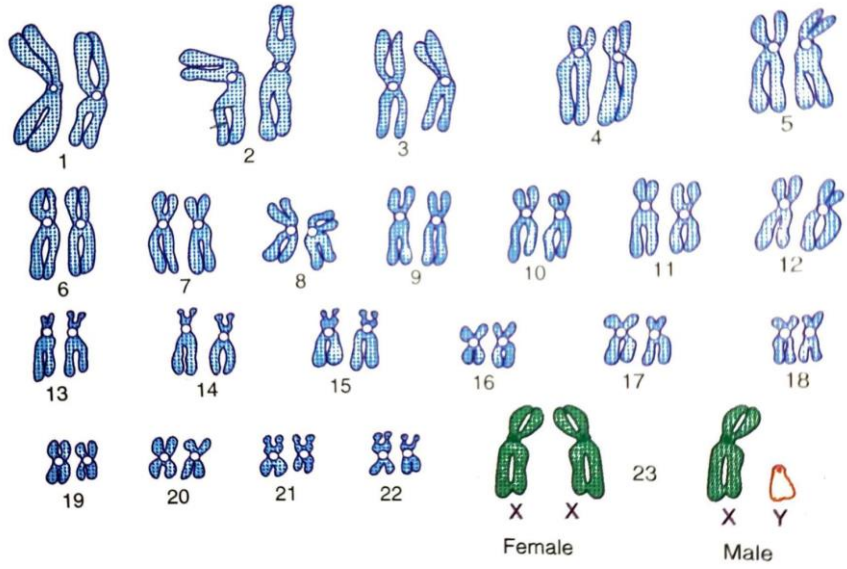
VARIATIONS IN POPULATION

Human beings as a species share many main characters or traits among themselves which identify the species Homo sapiens. Yet, the various races or tribes look different in several features. Further, within a family, members show differences in body features. These small differences among the individuals of the same species are called variations.

CHARACTER AND TRAITS: Any inheritable feature of an organism is a character. The alternative forms of a character are called traits. Ex:- Colour of eye is character Brown or blue is the trait.

CHROMOSOMES-THE CARRIERS OF HEREDITY

Photographs of the dividing cell nucleus can be taken through a high-power light microscope. These photographs are used for artificially arranging the chromosomes according to their size and shape on a chart (karyotype).



Chromosome number

The chromosome number is constant for the individuals of a species, and each body cell of that species has the same number of chromosomes. Humans have 46 chromosomes.

Chromosomes in Homologous Pairs

In each organism, the chromosomes occur in even numbers. This is so because they always occur in pairs. The two chromosomes of

each pair are similar in size and shape and are derived as one each from the two parents.

Homologous chromosomes

A pair of corresponding chromosomes of the same and size, one obtained from each parent.

THE TWO MAIN CATEGORIES OF CHROMOSOMES AUTOSOMES AND SEX CHROMOSOMES

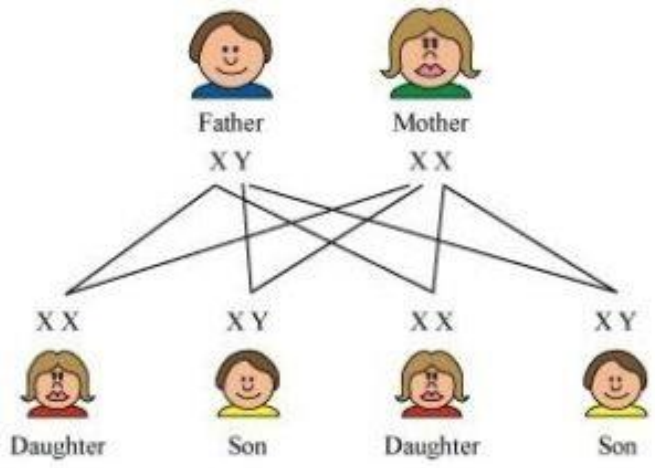
The chromosome pairs numbered 1-22, has identical chromosomes and these are categorised as autosomes. But the 23rd pair is different and its chromosomes are called sex chromosomes which are designated as X and Y.

The XX pair with similar partners is found in females whereas the XY pair with dissimilar partners is found in males.

Autosomes are the kind of chromosomes which determine general body features like complexion, height, seed colour, etc. Humans have 22 pairs of autosomes. While sex chromosomes (also called as allosomes) are the kind of chromosomes that determine the sex of an organism.

SEX DETERMINATION --- SON OR DAUGHTER

The sex of the child depends upon the kind of sperm that fertilises the egg. The egg contains only one X chromosome, but half of the sperms released into the genital tract of the female during coitus are X-bearing and the remaining half are Y-bearing.



CHROMOSOMES --- CARRIERS OF GENES

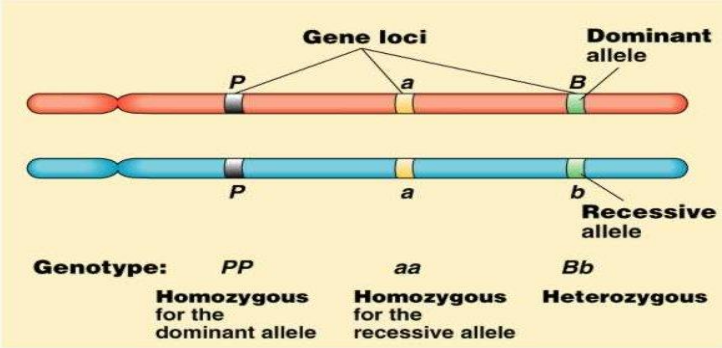
The characteristics of species including physical appearance, body functions, behaviour, etc., are not simply the outcome of chromosome number, but these are the result of the units called genes which the chromosomes carry.

The word "gene" was coined by geneticist Wilhelm Johanssen in 1909 to simply describe what parents passed to their offspring.

GENES AND THEIR ALLELES

The two alternative forms of genes are called the alleles.

ALLELES: Alternative forms of a gene occupying the same position (locus) on homologous chromosomes and affecting the same characteristic but in different ways.



Dominant Allele: The kind of allele which expresses itself regardless of the presence of another allele/s for a given gene. e.g. The dominant allele for height in garden pea is tall.

Recessive Allele: The kind of allele whose expression is suppressed in presence of a dominant allele for a given gene. A recessive allele can only express itself in presence of the same recessive allele. e.g. The recessive allele for height in garden pea is drawf.

GENOTYPE AND PHENOTYPE

The three situations pertaining to any pair of genes, as for example in tongue rolling, can be as follows:

(i) RR (both dominant) TONGUE ROLLER

(ii) Rr (one dominant, one recessive) TONGUE ROLLER

(iii) rr (both recessive) NON-ROLLER

The genetic constitutions (pertaining to the kinds of genes possessed) are called genotype and the expressed shown character is called phenotype.

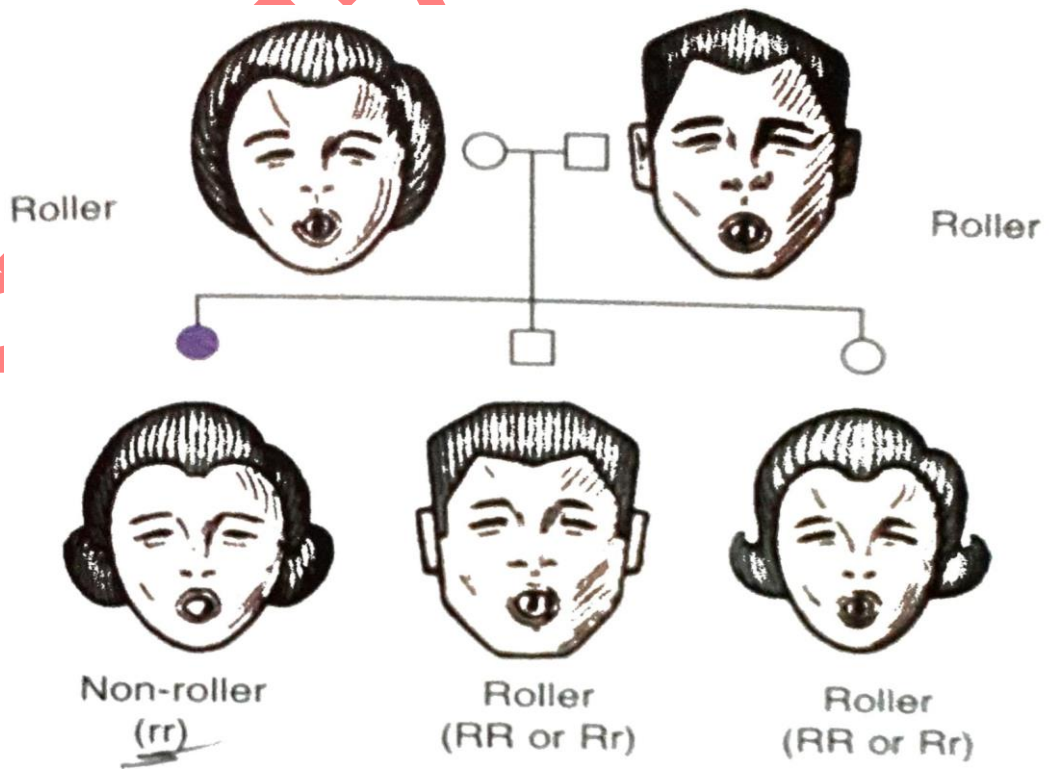
(i) homozygous dominant, that has similar pair RR (homo: similar, zygos: pair)

(ii) heterozygous dominant with dissimilar pair Rr (hetero: different, zygos: pair)

GENOTYPE AND PHENOTYPE

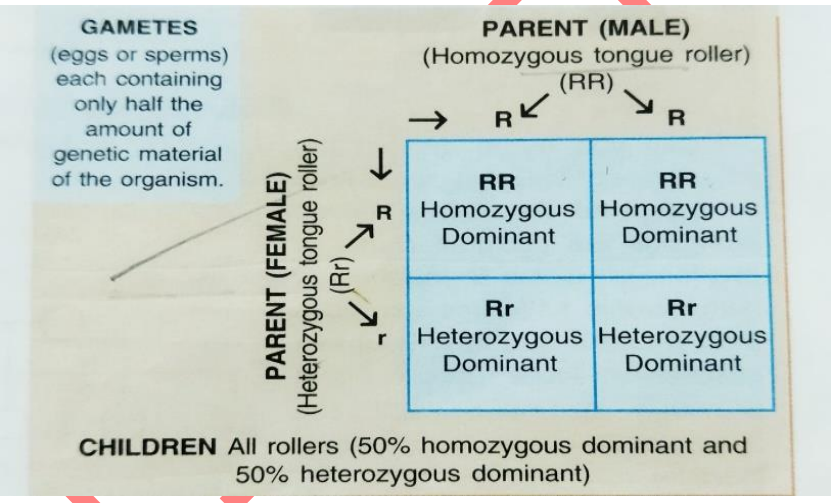
Genotype:- The set of genes present in the cells of an organism

Phenotype:- The observable characteristics which are genetically controlled.



FROM PARENTS TO CHILDREN --- TONGUE ROLLING --- AN EXAMPLE OF INHERITANCE - Pedigree Chart.

Punnett square: It is a simple diagram in which the different types of gametes (sex cells with the concerned trait) of one (female) parent are placed along one side of the square and those of the other parent (male) are placed along the other side. Then, the possible combinations (genotypes) of the opposite gametes are given in the sub-squares. The resulting phenotypes can be written under the genotypes.



SEX-LINKED INHERITANCE

Sex-linked inheritance is the appearance of a trait which is due to the presence of an allele exclusively either on the X chromosome or on the Y chromosome.

'X' linked inheritance:

Certain disorders caused due to heredity such as haemophilia and colour-blindness are more common in males than in females. Such defects are due to recessive genes, which occur on the 'X' chromosome! Colour blindess is an inherited disease due to which affected individuals cannot differentiate between certain colours, mostly red and green.

Haemophilia is a genetic disorder in which the sufferers (homozygous recessive female and the recessive X-bearing male) are at a risk of bleeding to death because the blood fails to clot in them. Rare cases of haemophiliac males do occur but practically none of haemophiliac females.

MENDEL'S EXPERIMENTS ON INHERITANCE

The basic principles of genetics were discovered for the first time by Gregor John Mendel in the mid- nineteenth century. Mendel was an Austrian monk and he conducted breeding experiments on garden pea (Pisum sativum) out of sheer interest. His findings became a milestone in biology.

Mendel had selected garden pea for three reasons:

1. Many varieties were available in alternative forms of a character.
2. Varieties were available in pure forms that bred true, i.e. produced the same type generation after generation.
3. Peas are normally self-pollinated but self-pollination could be prevented by removing corresponding reproductive parts (male part stamens and the female part carpels) of the flower and could as well be cross- pollinated artificially.

The two kinds of ratios in the two kinds of hybridisations are as follows:

Monohybrid ratios in F_2 generation:

Phenotypic ratio - 3:1

Genotypic ratio - 1:2:1

Dihybrid ratios in F_2 generation:

Phenotypic ratio - 9:3:3:1

Genotypic ratio - (very complex)

MENDEL'S LAWS OF INHERITANCE

Mendel's generalizations of the results of breeding experiments are summarised under three laws:

1. Law of Dominance: Out of a pair of contrasting characters present together, only one is able to express itself while the other remains suppressed. The one that expresses is the dominant character and the one unexpressed is the recessive.

2. Law of Segregation: The two members of a pair of factors separate during the formation of gametes. They do not blend but segregate or separate into different gametes. The gametes combine together by random fusion at the time of zygote formation.

3. Law of Independent Assortment: When there are two pairs of characters, the distribution of the alleles of one character into the gametes is independent of the distribution of the alleles of the other character.

Application of Mendel's laws

(i) A knowledge of the basic Mendelian principles gives us an idea about the new combinations in the progeny of hybrids and enables us to predict their frequency.

(ii) Such information is of great importance to both plant and animal breeders for producing better breeds.

(iii) New types of plants with new combinations of useful characters can be produced by hybridisation.

MUTATION

Mutation is a sudden change in one or more genes, or in the number or in the structure of chromosomes.

Mutation alters the hereditary material of an organism's cells and results in a change in certain characters or traits.

For example:

(i) Sickle cell anaemia is a blood disease caused by a gene mutation. The mutation causes change in the DNA resulting in the production of sickle-shaped RBCs.

(ii) Radioactive radiations also alter the gene structure and their effects can be seen generation after generation. An atomic explosion which had occurred during World War-II 1945 in Japan (Hiroshima, Nagasaki), had led to a number of deformities in the body of plants and animals which are still persisting.